Opis choroby *

Definicja

A rare arthrogryposis syndrome characterized by arthrogryposis multiplex congenita with contractures involving multiple joints of the upper and lower limbs, camptodactyly of fingers and toes, skeletal abnormalities such as scoliosis and pectus excavatum, as well as variable speech and motor delay and hypotonia. Facial dysmorphism includes long eyelashes, periorbital fullness, ptosis, epicanthal folds, high arched/cleft palate, and micrognathia.

Dane

Klasyfikacja
Choroba

Synonimy
MYBPC1-related autosomal recessive non-lethal AMC syndrome
MYBPC1-related autosomal recessive non-lethal AMC syndrome

Kod ORPHA
498693

Kod OMIM
-

Kod ICD10
Q74.3

Kod ICD11
-

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*Źródło

orphanet